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(54) Title: IDENTIFICATION OF SNPs ASSOCIATED WITH HYPERLIPIDEMIA, DYSLIPIDEMIA AND DEFECTIVE CARBOHYDRATE METABOLISM

(57) Abstract: The present invention relates to a nucleic acid molecule comprising a chromosomal region contributing to or indicative of hyperlipidemias and/or dyslipidemias or defective carbohydrate metabolism, wherein said nucleic acid molecule is selected from the group consisting of: (a) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO: 1, wherein said nucleic acid sequence has one or more mutations having an effect on USF1 function; (b) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO: 1, wherein said nucleic acid sequence is characterized by comprising a guanine or an adenine residue in position 3966 in intron 7 of the USF1 sequence; and/or (c) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO: 1, wherein said nucleic acid sequence is characterized by comprising a cytosine or a thymine residue in position 5205 in, exon 11 of the USF1 sequence; wherein said nucleic acid molecule extends, at a maximum, 50000 nucleotides over the 5' and/or 3' end of the nucleic acid molecule of SEQ ID NO: 1. The present invention further relates to a diagnostic composition comprising a nucleic acid molecule encoding USF1 or a fragment thereof, the nucleic acid molecule disclosed herein, the vector, the primer or primer pair of the present invention or an antibody specific for USF1. Finally, the present invention relates to the use of the nucleic acid molecule of the invention for the preparation of a pharmaceutical composition for the treatment of hyperlipidemia, dyslipidemia, coronary heart disease, type II diabetes, metabolic syndrome, hypertension or atherosclerosis.

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